

# Challenges representing complex and modern families and sensitive information

Robin L. Bennett, MS, CGC

Licensed Genetic Counselor

Clinical Associate Professor

Genetic Medicine Clinic

University of Washington Medical  
Center

Family History Tool Meeting

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# Objectives

- Review some of the complex relationships that are documented in family history
- Explore the balance of “social” relationships and biological relationships in family history
- Discuss some of the sensitive information that may be included in a pedigree

# 50 Facebook Gender Identities

- Agender
- Androgyne
- Androgynes
- Androgynous
- Bigender
- Cis
- Cis Female
- Cis Male
- Cis Man
- Cis Woman
- Cisgender
- Cisgender Female
- Cisgender Male
- Cisgender Man
- Cisgender Woman
- Female to Male
- FTM
- Gender Fluid
- Gender Nonconforming
- Gender Questioning
- Gender Variant
- Genderqueer
- Intersex
- Male to Female
- MTF
- Neither
- Neutrois
- Non-binary
- Other
- Pangender
- Trans
- Trans Female
- Trans Male
- Trans Man
- Trans Person
- Trans\*Female
- Trans\*Male
- Trans\*Man
- Trans\*Person
- Trans\*Woman
- Transexual
- Transexual Female
- Transexual Male
- Transexual Man
- Transexual Person
- Transexual Woman
- Transgender Female
- Transgender Person
- Transmasculine
- Two-spirit

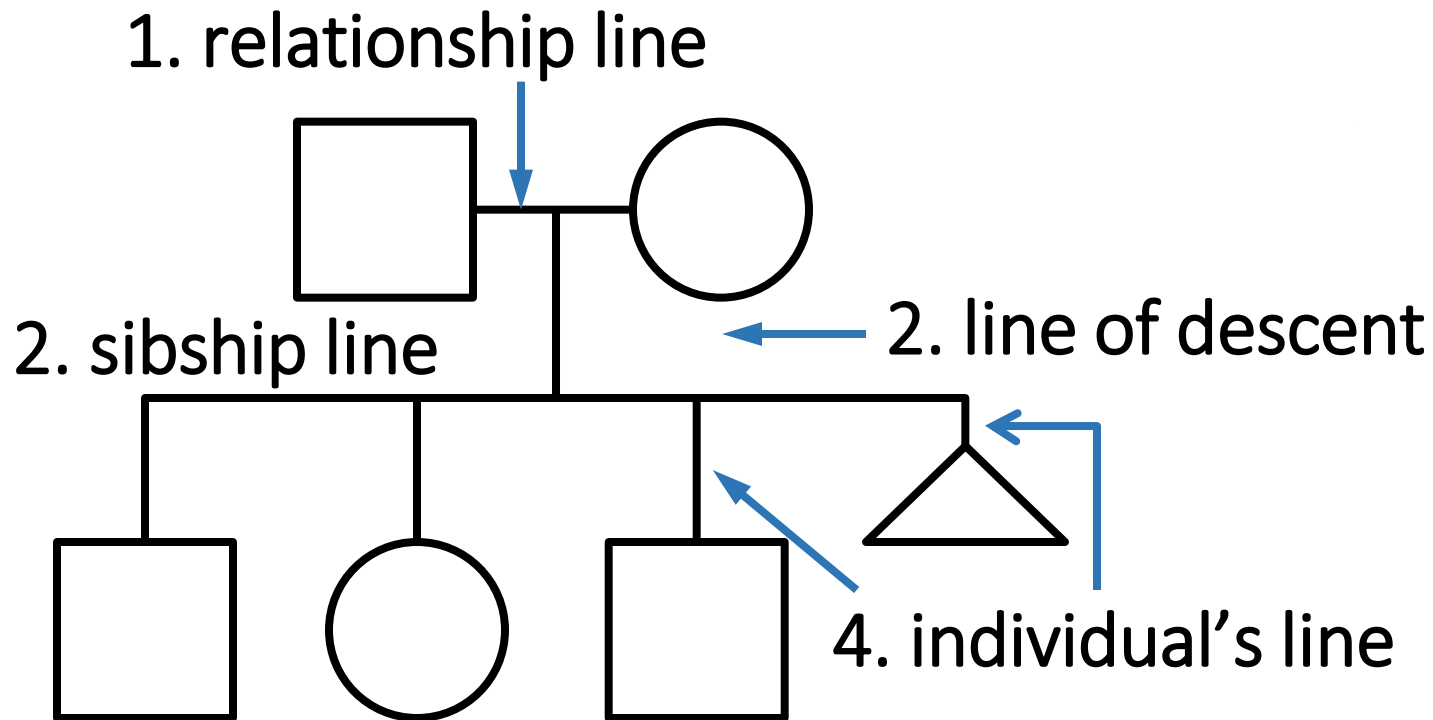
# Other Facebook Gender Identities

- Asexual
- Female to male trans man
- Female to male transgender man
- Female to male transsexual man
- F2M
- Gender neutral
- Hermaphrodite
- Intersex man
- Intersex person
- Intersex woman
- Male to female trans woman
- Male to female transgender woman
- Male to female transsexual woman
- Man
- M2F
- Polygender
- T\* man
- T\* woman
- Two\* person
- Two-spirit person
- Woman

# Documentation of family health history/pedigree

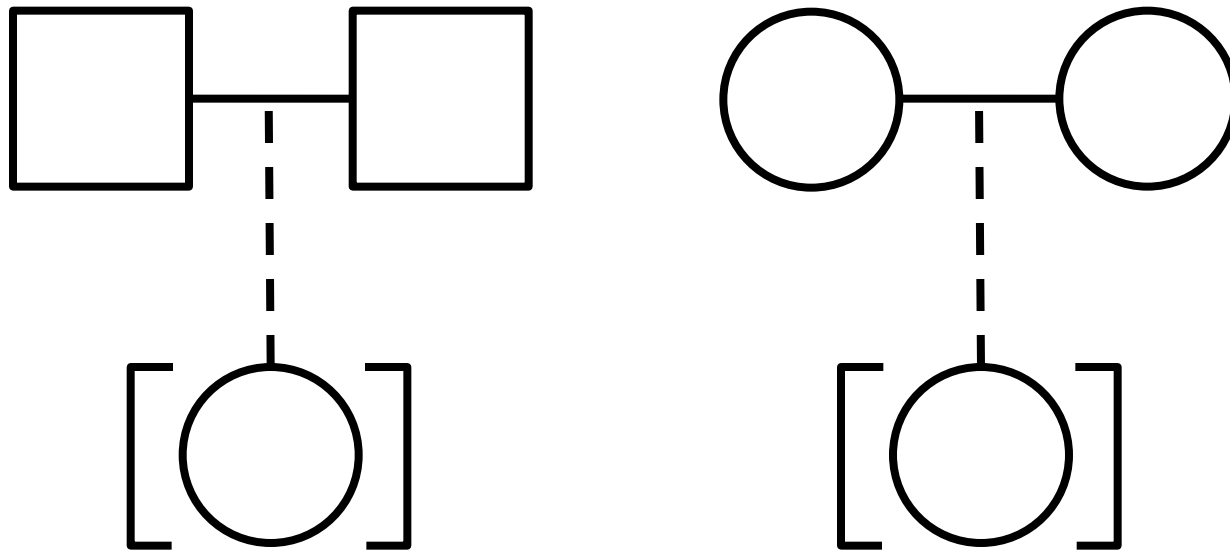
- Addressed in NSGC Standardized Pedigree Nomenclature
  - Bennett et al., *JGC*, 2008
- A diamond can be used to represent a transgender individual, or a person who does not identify as male or female
- Same sex partners can be noted on the pedigree
- Recommend that the male or female symbols (square/circle) define the phenotypic gender, with the gender at birth noted

# Pedigree line definitions



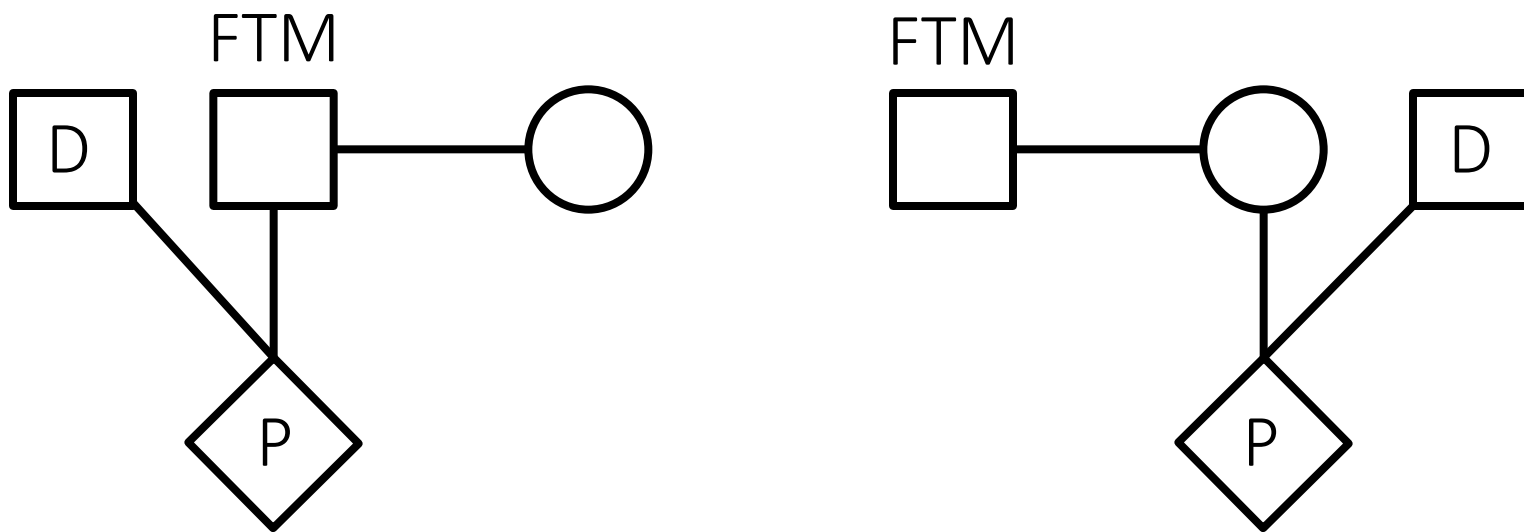
Bennett et al., *J Genet Couns* 2008

# Adoption



Adapted from Bennett et al., *J Genet Couns* 2008

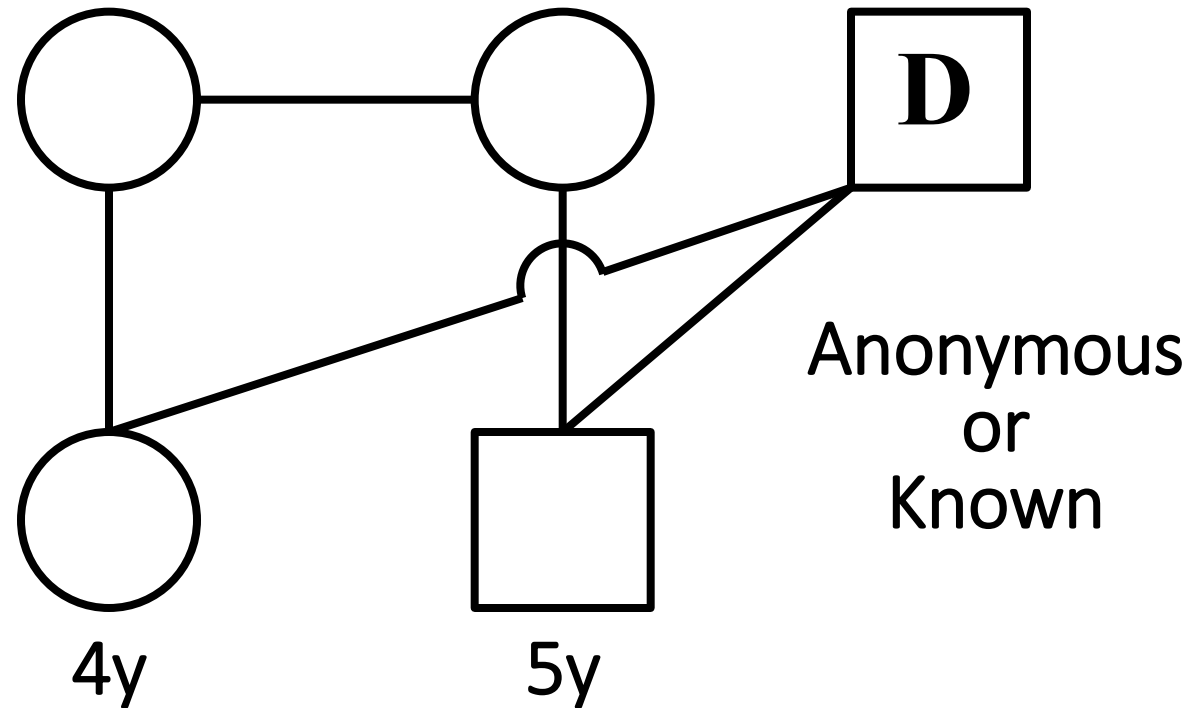
# Pedigree symbols for trans individuals



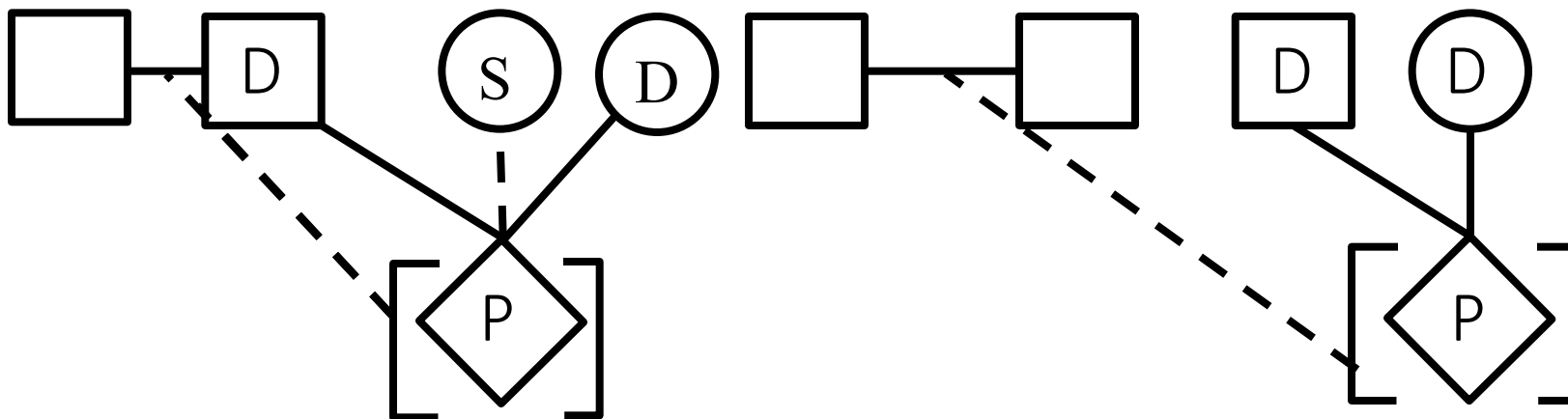
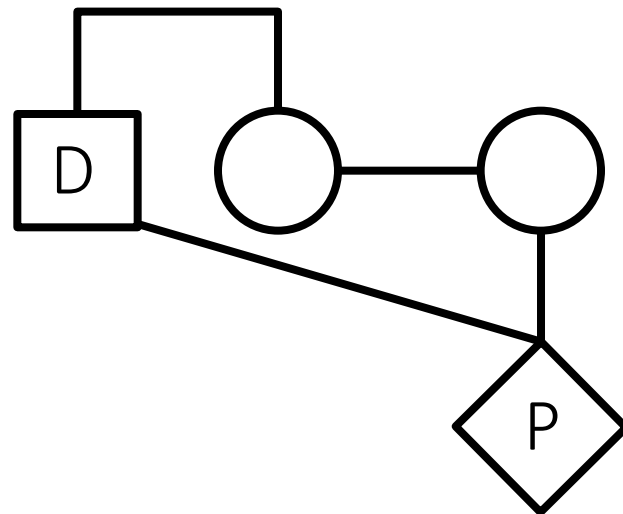
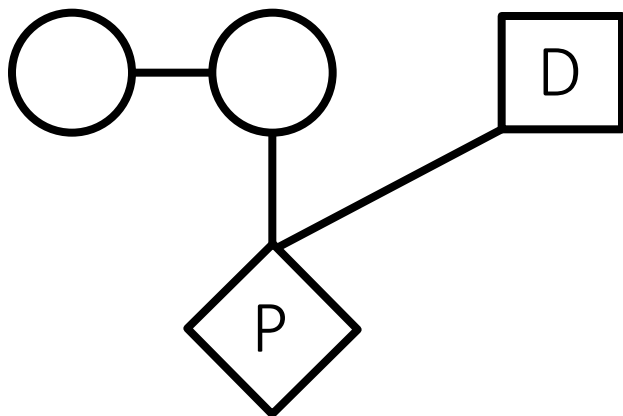
Adapted from Bennett et al., *J Genet Couns* 2008



## Example: Reciprocal IVF



## ART symbols



Adapted from Bennett et al., *J Genet Couns* 2008

# Pregnancies conceived with donor egg or sperm

- No accurate estimates of numbers, particularly for sperm donors
- Donor siblings registry (DSR) has 47,600 worldwide members
- Hundreds of sperm distribution facilities
- ~1/3 of donors donate to more than one clinic
- Most clinics request an updated family history
- FDA requires to keep donor health information for 10 years

# Sperm donation and collaborative arrangements with 1<sup>st</sup> degree relative

*Fertility & Sterility* 2012 98:797-803

Arrangement	Genetic & social relationship to offspring	Comments
Brother-to-brother	Paternal uncle is genetic father	Most common/acceptable
Brother-to-sister	Rearing mother is also genetic and gestational mother; social uncle is genetic father	Strong impression of incest but not illegal (neither sex or marriage involved); should be prohibited
Brother-to-sister (sister uses donated eggs)	Rearing mother is gestational mother, not genetically related to offspring; social uncle is genetic father; some cousins are half-siblings	Gametes not from consanguineous relationship but may create impression of incest

# Sperm donation and collaborative arrangements with 1<sup>st</sup> degree relatives

Arrangement	Genetic & social relationship to offspring	Comments
Father-to-son	Social paternal grandfather is genetic father; rearing father is genetic half brother	ASRM discourages donors >40 y because of concern for new mutations
Father-to daughter (daughter uses donated eggs)	Social maternal grandfather is genetic father	Divorced daughter who lived with father; Strong impression of incest
Son-to-father	Rearing father is genetic grandfather; offspring's social half-brother is genetic father, genetic paternal grandmother is rearing father's ex-wife	Usually second marriage for father (discouraged)

# Ovum donation and collaborative arrangements with 1<sup>st</sup> degree relatives

- Sister-to-sister
  - Social aunt is genetic mother
  - Some cousins are half siblings
- Other combinations possible but discouraged
  - Sister-to sister-in-law (brother's wife)
  - Daughter-to-mother
  - Mother-to-daughter

*Fertility & Sterility* 2012 98:797-803

# Surrogacy: traditional and gestational

- Sister-for-sister (traditional)
  - Social maternal aunt is genetic and gestational mother
  - Social cousins are half-siblings
- Sister-for-sister
  - Genetic relationships unchanged
  - Social maternal aunt is gestational mother
- Sister-for-brother
  - Genetic relationships unchanged
  - Social maternal grandmother is gestational mother
- Mother-for-daughter
  - Genetic relationships unchanged
  - Social maternal grandmother is gestational mother

# Frozen embryos and embryo adoption: Do we denote this differently on pedigree?

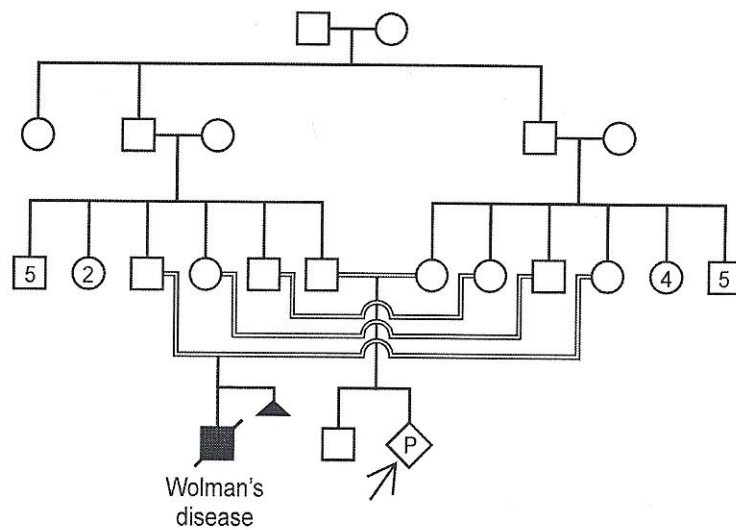
- Should we denote if “banked embryos” on pedigree
- Embryo adoption: Can be anonymous or from a relative or friend
- California Conceptions
  - Donor embryo
  - Conceived from donor egg and donor sperm



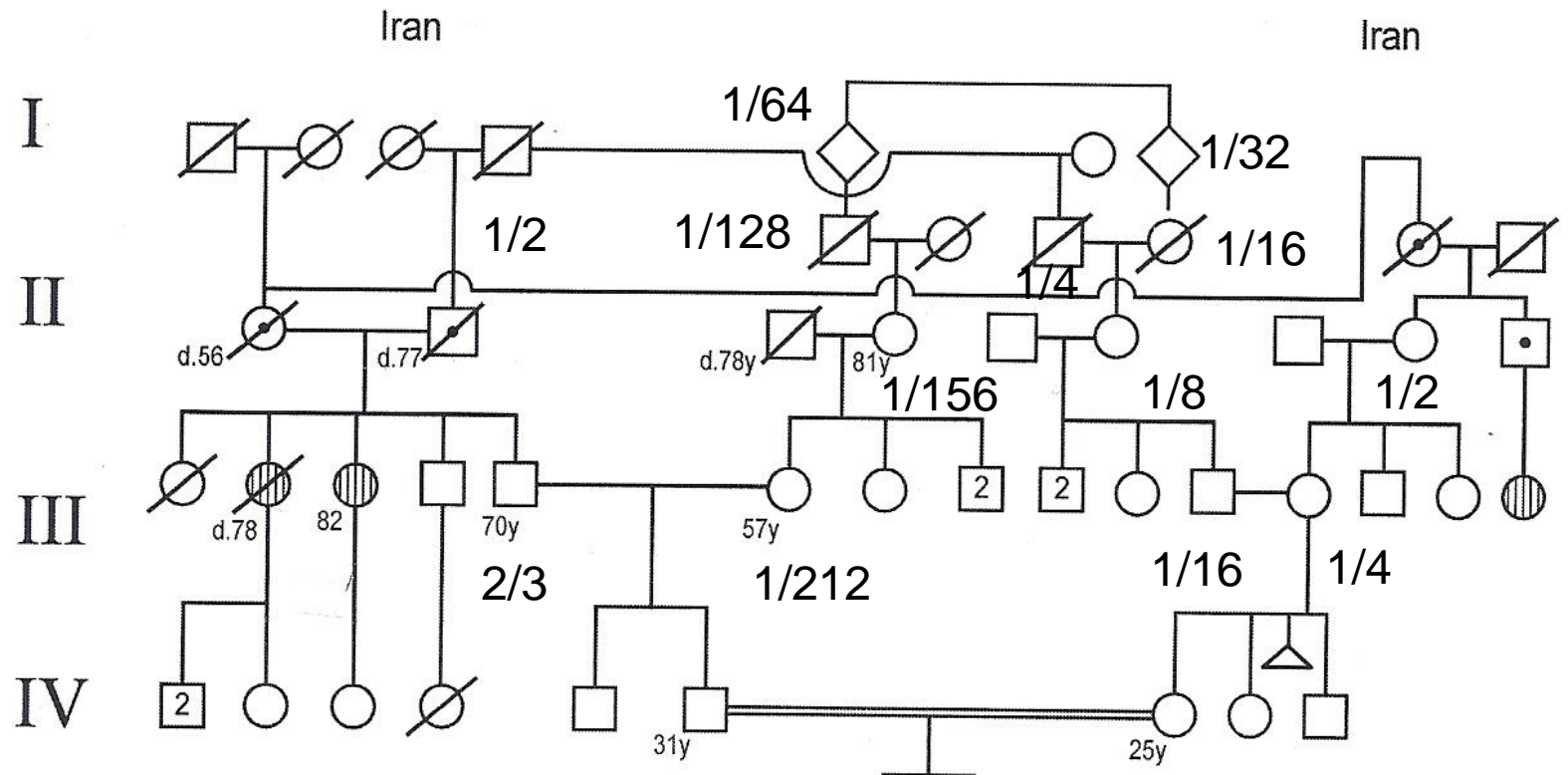
# Consanguinity

- No estimates of how common
- How far back do we go?
- How do we document multiple loops of consanguinity?
- What about genetic testing that suggests consanguinity?

# Afghanistan



# Multiple Loops of Consanguinity



$$(2/3 \times 2) + (1/212 \times 2) = 1/3 + 1/424 \approx 1/3 \quad (1/16 \times 2) + (1/4 \times 1/2) = 5/32$$

$$\text{Chance affected child: } 1/3 \times 5/32 \times 1/4 = 1/77$$

# Potentially Stigmatizing Information

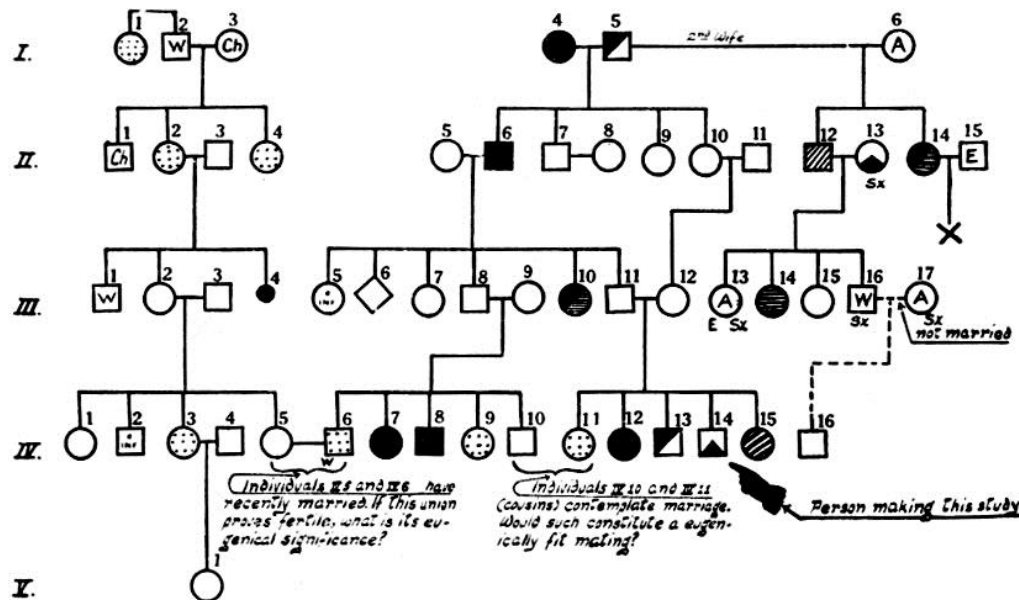
- Ages
- Cause of death
- Physical characteristics
- Genetic test results (asymptomatic)
- Mental illness/psychiatric history
- Suicide
- Substance abuse
- Pregnancy termination
- Misattributed paternity
- Infertility
- Pregnancies by gamete donation or surrogacy
- Adoption
- Ethnicity
- Consanguinity/incest

# **Pedigree Chart**

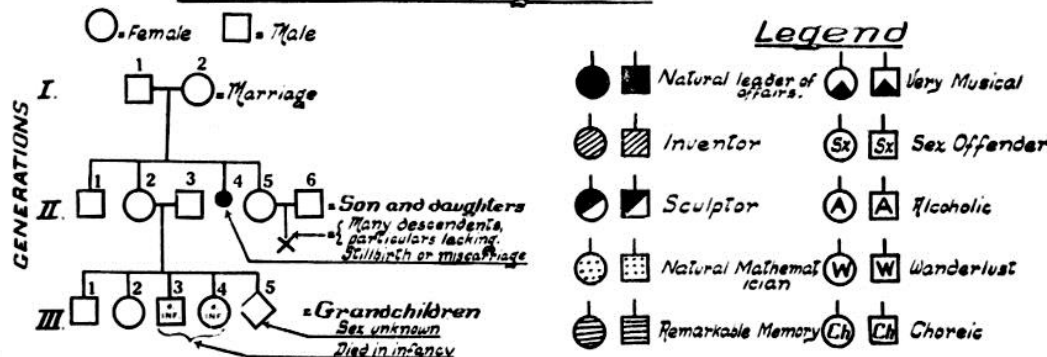
and for designating the family distribution of specific traits preparatory to making a descriptive analysis of the innate traits of each individual.

## **Small sample pedigree showing use of symbols, etc.**

Name of person making this study \_\_\_\_\_ Address \_\_\_\_\_ Date \_\_\_\_\_



### **Explanation of the above pedigree chart**



An explanation similar to this one should accompany each chart. The letters or symbols representing the traits characteristic of the family studied are arbitrarily chosen by the person making the chart. Only a few symbols and those for defects have been standardized—eg. I. Insane, F. Feeble-minded, Ch. Chorea, E. Epileptic, A. Alcoholic, etc. Note:—Such a chart is only the beginning of a eugenical study but it is very useful for showing graphically the family tree and for indicating in a general way the most striking traits of its various members. In order to determine the hereditary potentialities of an individual, each of the several members of his or her family network must be described—both as to good qualities and defects—with care, accuracy, and frankness. For this purpose the Individual Analysis Card is supplied.

**Eugenics Record Office,  
Cold Spring Harbor,  
Long Island, New York**